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Micromelic Dwarfism With Cone Epiphyses, Metaphyseal Dysplasia, and Vertebral Segmentation Defects

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We present the clinical and radiological findings in a newborn male with severe micromelic dwarfism, short neck, short and narrow upper thorax, and brachydactyly. At the age of 1 year mental development is slightly retarded. The X-ray findings of severe vertebral segmentation defects and a generalized metaphyseal skeletal dysplasia did not lead to a final, conclusive diagnosis. The present patient may be the first example of a new type of micromelic spondylo-epi-metaphyseal dysplasias.

KEY WORDS: vertebral segmentation, cone epiphyses, SEMD, SMD, short-limbed dwarfism

INTRODUCTION

The spondylo-epi-metaphyseal dysplasias (SEMD) constitute a heterogeneous group of skeletal dysplasias with great variety in severity of metaphyseal, epiphyseal, and vertebral involvement [Maroteaux, 1995; Vichi et al., 1990]. Whereas clinical and radiological findings may be extremely variable, hands and feet are usually not involved and the spine presents only mild anomalies.

We present a male child with micromelic dwarfism, short neck, narrow thorax, brachydactyly, and severe vertebral segmentation defects as a possible first example of a new type of spondylo-epi-metaphyseal dysplasia.

CLINICAL REPORT

The index patient of this report is a male newborn, second child of healthy, non-consanguineous parents with unremarkable family histories. The first child, a 2-year-old daughter, is normal. The father is 28 and the mother 36 years old. Pregnancy and delivery, at 41 weeks, were normal. Birthweight was 3,600 g, length 41.5 cm, and head circumference (OFC) 36 cm. No perinatal problems were noted, but clinical examination showed severe, short-limb dwarfism with predominant rhizomelic micromelia (Fig. 1). The neck was short, and the thorax short and narrow in its upper part. The head was relatively large with flat occiput, small mouth, and anteverted nares. The anterior fontanelle was normal. Abundant lanugo was present on both earlobes and presacral region. Hands and feet were small with both thumbs in fixed adduction and talipes equinovari. Genital examination showed a small phallus with narrow prepuce, and a normal scrotum with both testes in normal position. Neurological evaluation was normal. Findings on cardiac and abdominal echographies, and ophthalmological examination, were normal. Results of routine hematological, metabolic, and biochemical screening, including calcium, phosphorus, and alkaline phosphatases, were normal. A skeletal survey (Fig. 2) showed severe vertebral anomalies with multiple cervical fusion defects, thoracic hemivertebrae (D1, D3, D6), marked shortness of the long tubular bones with broad metaphyseal ends. No major problems were noted in the first months of life. At 4 months, length, weight, and OFC were 51 cm, 4,900 g, and 43.5 cm, respectively. At 9 months (length 58 cm, weight 6,660 g, OFC 47.7 cm) a skeletal survey confirmed the presence of multiple cervical fusions (posterior fusion C2-C3, anterior fusion C4-C5-C6; hemivertebrae D1, D3, D6 with discrete dextroconvex scoliosis; irregular structure of all thoracic and lumbar vertebrae with persistent coronal and sagittal clefting. Metacarpals, proximal, and median phalanges were short with broad, irregular metaphyses (Fig. 3) and the distal phalanges small with distal expansion. The halluces were broad and short with broad and irregular metaphyses. The median and distal phalanges of toes II to V were rudimentary. The long tubular bones of upper and lower limbs were short with broad, irregular metaphyses; there was marked cupping of the distal metaphyses of both femora (Fig. 4) with invaginated, small epiphyseal centers (cone epiphyses). The pelvis was small with hypoplastic ilia and

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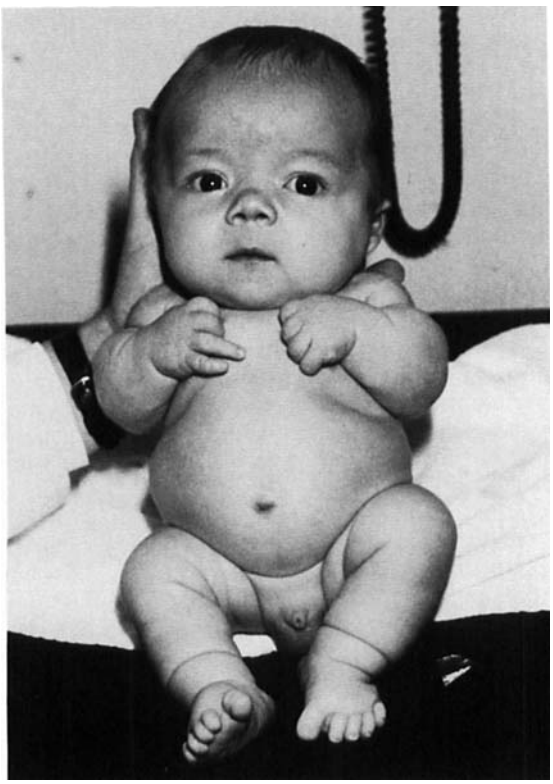

Fig. 1. General appearance.

absence of proximal femoral epiphyseal ossification. At 1 year, weight was 7,170 g, length 59.5 cm, and OFC 48.8 cm. Clinical findings corresponded well with those in the neonatal period. Motor development was severely retarded, and the boy could not sit or hold up his head without support. Mental development was slightly retarded with developmental level of 9 months on the Bayley Mental Developmental scale. Social contact was satisfactory.

Now, at 18 months (weight 7,950 g, length 65 cm, OFC 50.8 cm) he starts to sit without support but his head control is still poor. Mental developmental level on the Bayley scale is 12 months. Clinical findings are identical and X-ray films of the cervico-thoracic region confirm the cervical and upper dorsal fusion and segmentation defects (Fig. 5). Proximal femoral ossification centers are absent and the X-ray findings of the left knee are identical to the findings at 8 months with marked cupping of the distal femoral metaphysis with invagination of a small epiphysis (cone epiphysis—ball in cup configuration).

DISCUSSION

In this boy with a severe type of micromelic dwarfism clinical follow-up up to the age of 1 year showed a small number of associated findings: a characteristic cranio-

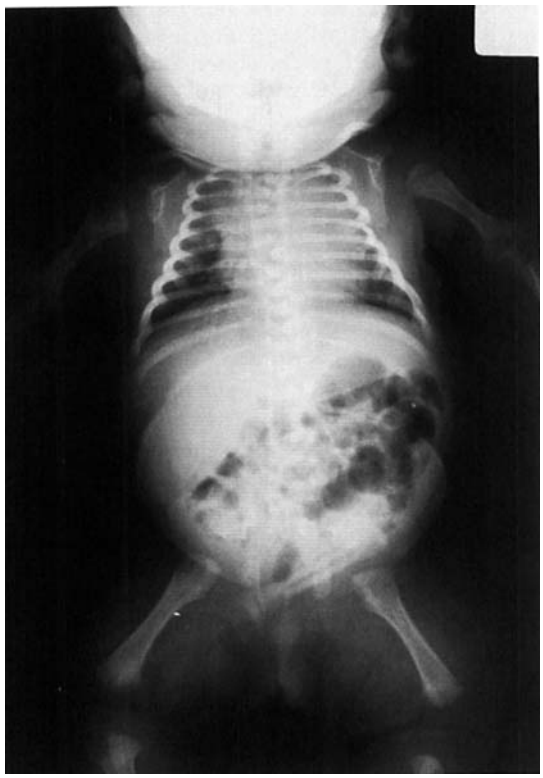

Fig. 2. X-ray skeletal survey in the neonatal period.

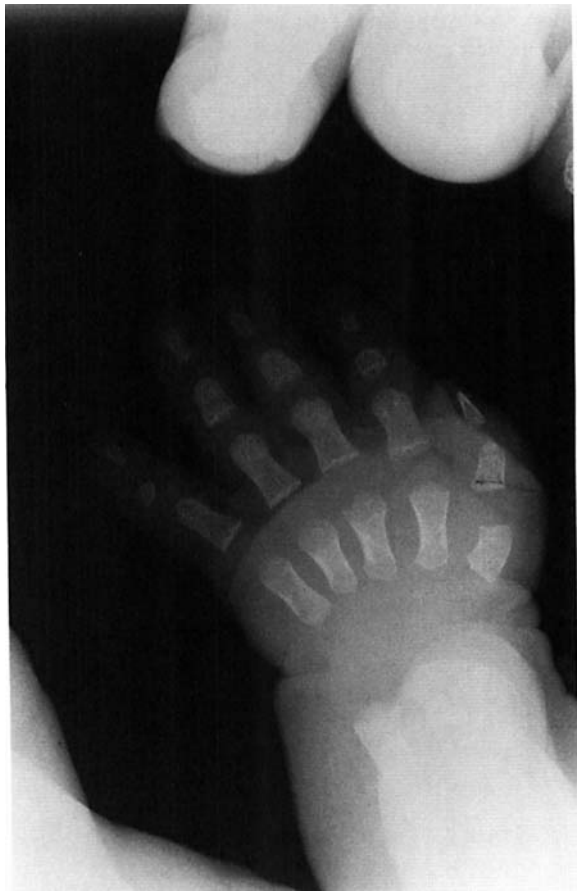

Fig. 3. X-ray view of the left hand (age 9 months).

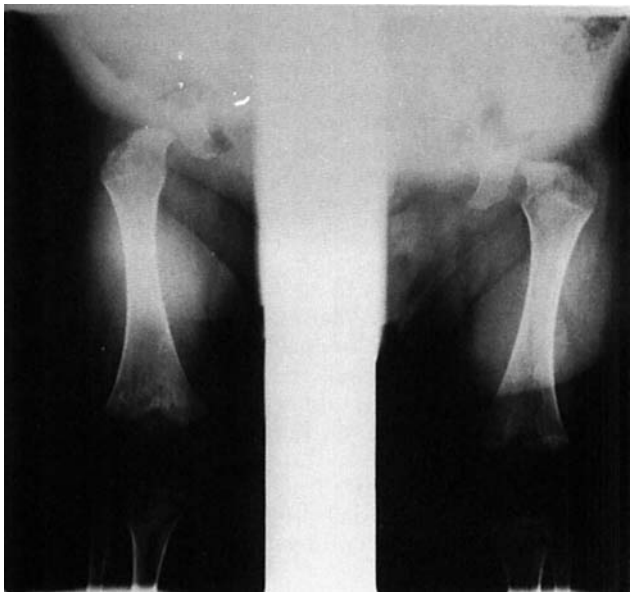

Fig. 4. X-ray view of femora (age 9 months).

facial appearance with relatively large head; depressed nasal bridge; small nose with anteverted nares, long, fine philtrum, and small mouth with fine lips; short neck; abundant lanugo on the ears and the lumbosacral region; a small phallus with contrasting normal scrotum and testes; and slightly retarded mental development contrasting with severe delay in motor development. Skeletal roentgenograms showed severe vertebral anomalies with cervical segmentation defects and multiple thoracic hemivertebrae, and distinct metaphyseal changes with broad metaphyses of the short and long tubular bones. The metaphyseal abnormalities were more evident on the skeletal films at 9 months and the distal metaphyses of both femora showed irregularity and distinct cupping with invagination, cone-shaping of the small epiphyseal centers.

Fig. 5. X-ray view of the cervicothoracal vertebral column at the age of 18 months.

The simultaneous occurrence of involvement of spine, metaphyses, and epiphyses may categorize this observation in the complex and heterogeneous group of the spondylo-epi-metaphyseal dysplasias (SEMD), but an extensive search of reported forms gave no conclusive suggestions for a final diagnosis [Azouz, 1987; Bueno et al., 1984; Freisinger et al., 1993; Kozlowski, 1974; Maroteaux, 1995; Verloes et al., 1991; Vichi et al., 1990].

In the micromelic type of SEMD (also reported as "enchodromatosis-vertebral anomalies") [Kozlowski, 1974] rhizomelic micromelia is associated with epiphysic-metaphyseal dysplasia and mild scoliosis, but there is only mild involvement of the spine with hypoplasia of thoracic vertebral bodies. In this condition hands and feet are long, facial appearance is "coarse" and the anterior fontanelle large. Congenital SEMD [Maroteaux, 1995] is characterized by short neck, rhizomelic micromelia, flattening of vertebral bodies, scoliosis, pelvic dysplasia, and underossification of the femoral head and neck. Hands and feet are normal and severe segmentation defects of the spine are absent. Vichi et al. [1990] reported an atypical variant of SEMD with involvement of hands and feet, but neither in this form vertebral defects are present. The association of severe dwarfism with short neck, short and narrow thorax, rhizomelic micromelia with metaphyseal widening, brachymetacarpy, and adducted thumbs could suggest the diagnosis of dyssegmental dysplasia [Maroteaux, 1995], but the presence of vertebral segmentation defects and epiphyseal abnormalities excludes this diagnosis. Cone-shaped distal femoral epiphyses together with involvement of hands and feet are found in acroscyphodysplasia [Verloes et al., 1991], but also this diagnosis can be excluded by the presence of severe involvement of the spine. Hemivertebrae and rhizomelic micromelia are typical findings in dysspondylo-chondromatosis [Freisinger et al., 1993], but in this condition epiphyses, hands, and feet are only mildly involved and unequal length of the limbs is a diagnostic sign.

To summarize, the clinical and radiological findings in the present boy may be the first example of a new type of spondylo-epiphyseal-metaphyseal dysplasia but further observations are needed to confirm this suggestion and to further delineate the spectrum of clinical and radiological findings of this condition.

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